Radiology Basics, Part V: Congenital Anomalies of the Gastrointestinal Tract

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The Stomach
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CONGENITAL ANOMALIES OF THE GASTROINTESTINAL (GI) tract may involve any part of the primitive tube from the hypopharynx to the anus. Stenoses, atresias, duplications, and obstructions are among the common lesions of the gastrointestinal tract.1,2 These lesions can be differentiated on the basis of data from the history along with physical examination, clinical presentation, and radiographic imaging. Findings from the history and physical examination significant in making the diagnosis of GI obstruction may include: (1) polyhydramnios, (2) large gastric aspirates, (3) abdominal distention, (4) bilious or nonbilious emesis, and (5) failure to stool.1

The gastrointestinal tract can be divided into the stomach, the small bowel, and the large bowel. The small bowel consists of three portions: the duodenum, the jejunum, and the ileum. The large bowel consists of the cecum, the colon, the rectum, and the anal canal. The orientation of the gastrointestinal anatomy is outlined in Figure 1.

Valuable information regarding the location of the congenital lesion may be obtained from radiographic imaging. Under normal circumstances, air is seen in the stomach immediately after birth. Within approximately one hour, the proximal portion of the small bowel and segments of the colon will be air filled. Within three hours, the distal portion of the colon will be air filled.2,3 Remember, however, that many parts of the gut are normally invisible on x-ray because they contain fluid or feces or are collapsed.

Gastric outlet obstruction can be the result of events, such as in utero vascular accidents, that occur during the embryologic development of the GI tract.4 When there is an obstructive lesion, distention of the intestine proximal to the obstruction will be noted on the x-ray. A flat plate film, sometimes referred to as a plain film or KUB (kidney, ureter, and bladder), is most commonly used to identify GI abnormalities. A lateral film, which is helpful in identifying free air or fluid, may also be obtained. In addition to the flat plate film, x-ray studies using contrast material are instrumental in identifying the location of many obstructions.

For the clinician interpreting an abdominal x-ray, the following step-by-step approach may be helpful so that subtle changes are not overlooked:
1. Identify the level of the diaphragm. In the absence of pulmonary disease the diaphragm should be at the ninth thoracic vertebrae during the inspiratory phase of respiration. This may be elevated in cases of obstruction with distention or free air in the abdomen. Look at the bony
structures, which include the vertebral column, lower ribs, and pelvis. Note any asymmetry. The placement of catheters, if any, in abdominal vessels should be noted.

2. Next examine the soft tissues of the left upper quadrant, right upper quadrant, both flanks (the outer lateral areas of the abdomen), mid-abdomen, and pelvis. In each quadrant, check for normal organ masses (i.e., liver, spleen, and stomach). Any shift of organs or intestine from the normal location should be noted.

3. Assess the contents of the GI tract. Look at the abdominal contour for bulging flanks or mild to marked distention. Distention may be due to physiologic or structural obstruction or accompany respiratory distress or mechanical ventilation. Follow the course of air throughout the GI tract, keeping in mind the age of the patient and the normal progression of air through the GI tract postnatally. A complete obstruction will cause opacity distal to the obstruction. Look for free air. A large collection of free air will lead to abdominal distention and an elevated diaphragm, and outlines structures such as the falciform ligament. Small amounts of free air may be evident on either side of the intestinal wall (i.e., “double wall sign”): The intestine is outlined by air in the peritoneum and air in the intestinal lumen. This is best appreciated on the lateral film.

4. Evaluate for the presence of radiodensities such as calcifications and air fluid levels. If these exist, calcifications within or around the bowel will be seen as white masses and a bowel filled with fluid will cast vague and confluent gray shadows. Calcifications may be caused by sterile meconium peritonitis, which appears as small, rounded opacities; renal calculi; teratomas that are retroperitoneal; or neuroblastoma.

DEVELOPMENT OF THE STOMACH

Development of the gastrointestinal tract, an early event, begins during the fourth week of gestation and is completed by the tenth week. The stomach arises during the fourth week of gestation as a spindle-shaped dilation of the foregut. In the subsequent weeks, the stomach dilates and rotates, resulting in a change in appearance and position. The rotation is around a longitudinal and anteroposterior axis. The effects of this longitudinal rotation are (1) the lesser curvature moves to the right, (2) the greater curvature moves to the left, (3) the original left side becomes the ventral surface, and (4) the original right side becomes the dorsal surface. During the anteroposterior rotation, the caudal (pyloric) portion moves to the right and upward, whereas the cephalic (cardiac) portion moves left and downward.
Because the embryologic development of the stomach is relatively simple, congenital anomalies, such as pyloric atresia, are rare. Anomalies may range from stenosis or atresia of the pylorus of the stomach, microgastria or hypoplastic stomach, volvulus of the stomach, to duplication cysts which can simulate pyloric atresia.

**ANOMALY OF THE STOMACH: PYLORIC ATRESIA**

Pyloric atresia is a complete obstruction of the gastric outlet—the pylorus. This congenital anomaly represents less than 1 percent of all atresias of the GI tract.\(^9,10\) Although it is usually a solitary anomaly, it has been described in association with multiple atresias of the small and large bowel.\(^10\) An association between pyloric atresia and epidermolysis bullosa has also been well recognized by clinicians.\(^10\)

**Clinical Features**

Pyloric atresia is characterized by polyhydramnios. Presenting symptoms at birth are nonbilious emesis, abdominal distention, and, possibly, visible peristalsis noted in the upper abdominal quadrants.\(^5,11\) Rarely, further examination may reveal evidence of skin lesions that are consistent with epidermolysis bullosa.

**Radiographic Imaging**

In the presence of pyloric atresia, an abdominal film shows air confined to the stomach. The characteristic sign on the film is a single gastric bubble (Figure 2). If calcifications are noted in the abdominal area, multiple atresias are likely to be present along with atresia of the pylorus.

**CASE STUDY**

Baby Girl P was a 1,460 gm infant born at 32 weeks gestation by spontaneous vaginal delivery to a 27-year-old Hispanic mother who was gravida 5, para 1, ab 1, newborn death 3. Maternal history was as follows: blood type O positive, VDRL negative, rubella immune, tuberculin purified protein derivative (PPD) positive, chest x-ray normal, and HbsAg negative. The pregnancy was complicated by premature rupture of membranes (PROM) one week prior to delivery and preterm labor. Antibiotic therapy was given for treatment of the PROM. The mother also received steroids in anticipation of the delivery of a preterm infant. Tocolytic magnesium sulfate therapy failed, and labor progressed. The infant was delivered vaginally with Apgar scores of 6 and 8 at one and five minutes. Resuscitation consisted of suctioning of the thin meconium noted at delivery, oxygen administration, drying, and stimulation.

Baby Girl P was transferred to the NICU. Upon physical examination, skin bullae and sloughing of the skin were noted, as well as a small protruding mass in the left upper abdominal quadrant and nonbilious emesis. The findings of the abdominal x-ray taken at 24 hours of age are shown in Figure 3, and those at 36 hours of age are shown in Figure 4. A skin biopsy was obtained and confirmed the diagnosis of epidermolysis bullosa.
X-Ray Evaluation (Figure 4)

**Indication** for the x-ray was the presence of an abdominal mass and nonbilious emesis.

**Penetration** appears to be normal.

**Rotation** appears to be normal.

The soft tissues of the neck, chest, and extremities are of normal thickness.

The bony framework is intact, with 12 ribs bilaterally and normal vertebrae.

The chest shows clear lung fields, the presence of air bronchograms, the diaphragm at T10 bilaterally, normal heart size.

In the abdomen, the stomach is on the left and is dilated, with no air distal to the stomach. The tip of the gastric tube is located in the stomach. The remainder of the intestinal tract shows lack of air throughout.

The impression is that the history, clinical presentation, and x-ray findings are consistent with a diagnosis of pyloric obstruction.

The diagnosis of pyloric atresia was confirmed by the surgical team during the exploratory laparotomy. The atretic area was resected and a gastrostomy tube was placed. Baby Girl P died at one month of age as a result of complications of epidermolysis bullosa.

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**The Small Bowel**

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**Congenital Obstruction of the Small Bowel** could involve the duodenum, jejunum, or ileum. The infant usually presents with bilious emesis, abdominal distention, and failure to pass meconium stool. To determine the location of the obstruction, plain and contrast radiographic studies are done. The following steps are recommended when evaluating the radiographic studies:

1. Identify the level of the diaphragm, which may be elevated with intestinal obstruction. Next look at the bony structures, noting any asymmetry.
2. Evaluate the soft tissues in each of the four quadrants and in the pelvis.
3. Assess the contents of the gastrointestinal tract. Look at the abdominal contour for evidence of distention. Follow the course of the air throughout the G1 tract. The mucosal pattern is not well defined in infants. This is an important point to keep in mind when evaluating intestinal films to avoid overdiagnosing intestinal abnormalities.

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**TABLE 1**

<table>
<thead>
<tr>
<th>Factors Associated with Duodenal Obstruction</th>
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<tr>
<td>Down syndrome</td>
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**Small Bowel Development**

The small intestine is formed by the terminal portion of the foregut and the cephalic portion of the midgut. There is rapid elongation of the gut, which quickly exceeds the growth of the rest of the body, including the abdominal cavity. As a result, the small intestine herniates into the extraembryonic coelom of the proximal umbilical cord. This physiologic herniation begins at the sixth week of gestation, with return of the gut to the abdominal cavity during the tenth week. Development of the small intestine occurs in four steps: (1) herniation, (2) rotation, (3) retraction, and (4) fixation. Herniation is characterized by rapid elongation and by formation of U-shaped loops, which protrude into the umbilical cord. A 270-degree rotation in a counterclockwise pattern begins with a 90-degree turn of the intestine while it is still positioned in the umbilical cord.

When the intestine returns to the abdominal cavity, an additional 180-degree turn takes place. The lumen of the intestine becomes obliterated temporarily by rapid epithelial growth, but later, recanalization occurs. Retraction, or re-entry of the gut, occurs during the tenth week of gestation. During the final step of development, fixation occurs. Abnormalities can occur due to failure of recanalization, rotation, or fixation. Small bowel atresia or stenosis may develop from failure of recanalization. Malrotation and volvulus could occur if the gut does not properly rotate and fix within the abdominal cavity.

**Small Bowel Anomalies: Duodenal Obstructions**

Complete or partial blockage of the duodenum can cause duodenal obstruction. The most common site of obstruction is at, or just below, the entrance from the stomach to the

**TABLE 2**

<table>
<thead>
<tr>
<th>Clinical Manifestations of Duodenal Obstruction</th>
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<tr>
<td>Abdominal distention (may or may not be present; if present, left upper quadrant only)</td>
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<tr>
<td>Possible passage of meconium, within first 24 hours only</td>
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<tr>
<td>Dehydration</td>
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<td>Indirect hyperbilirubinemia</td>
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duodenum, the ampulla of Vater.\textsuperscript{13–15} Duodenal atresia, in which the lumen of the GI tract is disrupted, is a complete blockage, whereas duodenal stenosis, a narrowing of the lumen, may result in a partial blockage.\textsuperscript{14}

Duodenal obstruction may result from the failure of recanalization of the duodenal lumen (most common) or from an intrauterine vascular ischemia.\textsuperscript{13–15} Obstructions are categorized as intrinsic or extrinsic in nature. Intrinsic obstructions are located within the intestinal wall and include duodenal atresia, stenosis, webs, and duplications. Intrinsic obstructions have a two-thirds greater incidence than extrinsic obstructions. Extrinsic compression can be caused by an annular pancreas, Ladd’s bands, mesenteric artery syndrome, malrotation, or volvulus.\textsuperscript{16,17}

Normal positioning of the small bowel is the characteristic C-contour, and the duodenum is fixed to the left of midline at the ligament of Treitz. In malrotation, the positioning of the small bowel is predominantly to the right and the colon to the left.

Abnormal rotation of the duodenum and small bowel results in the formation of peritoneal bands (Ladd’s bands), which can lead to duodenal obstruction. When volvulus occurs, the intestine has twisted around the superior mesenteric artery, resulting in vascular compromise.

Annular pancreas results from the failure of the pancreatic tissue to properly rotate around the duodenum, resulting in the formation of a ring of pancreatic tissue encircling the duodenum. It can cause either a partial or complete obstruction of the duodenum.

Duodenal obstruction has a high association with other congenital anomalies, as shown in Table 1.

**Clinical Features**

There is typically a history of polyhydramnios in infants having a high GI tract obstruction.\textsuperscript{13–17} Generally, infants with duodenal obstruction present with bilious emesis and/or bilious gastric aspirate >25 ml shortly after birth.\textsuperscript{13–17} Other clinical manifestations are shown in Table 2.

**Radiographic Imaging**

Once the diagnosis of intestinal obstruction is considered, a plain radiograph of the abdomen should be obtained. With duodenal obstruction, the typical “double bubble” sign is present (Figure 5). This sign documents the presence of air in the stomach and first segment of the duodenum.\textsuperscript{13,14,18}

If no air is seen distal to the duodenum, the infant probably has an atresia. If a few isolated areas of air are seen past the duodenum, this may represent duodenal stenosis, although further contrast studies are indicated to rule out malrotation with or without volvulus and other obstructions such as annular pancreas.\textsuperscript{13,14,18} See Figures 6 and 7.

In malrotation without volvulus, there is displacement of the cecum and the terminal ileum upwardly and medially. If malrotation complicated with volvulus is suspected, the contrast studies reveal a tapering deformity of obstructed duodenum. Another radiographic finding seen with volvulus is the absence of the ligament of Treitz. Normally, the ligament of Treitz is located to the left of the spine.

**Case Study**

Baby Girl B, a 37 week, 2,560 gm female, was born by cesarean section, secondary to fetal distress, to a 24-year-old gravida 4, para 2 mother with good prenatal care. The pregnancy was uncomplicated, with maternal history as follows: blood type AB positive, VDRL negative, rubella status immune, and HbsAg negative. A fetal ultrasound during the second trimester indicated polyhydramnios, with markedly dilated stomach and a second fluid-filled structure. Spontaneous rupture of membranes, with clear fluid, occurred 30 minutes prior to delivery. Abruptio placentae was noted on delivery. Upon delivery, the neonate was depressed, requiring bag-and-mask ventilation for 10–15 seconds. Apgar scores were 6 and 8 at one and five minutes, respectively. An orogastric tube was inserted, and 40 ml of bilious fluid were aspirated. Baby Girl B was taken to the NICU for further evaluation and management. Figure 8 shows the first KUB film for this patient.

**X-Ray Evaluation (Figure 8)**

**Indication** for the x-ray was prenatal suspicion of GI obstruction and large gastric aspirate at birth.

**Penetration** appears to be normal.

**Rotation** is slightly to the left.

The **soft tissue** of the neck is not visible. The soft tissues of the chest and extremities are of normal thickness.

The **body framework** is intact, with 12 ribs bilaterally and normal vertebrae.

The **diaphragm** is at T10 on the right and at T10–T11 on the left.
Abdomen: The stomach, on the left, has been somewhat decompressed. The proximal duodenum is markedly dilated, with no air seen in the remainder of the small bowel or in the colon. A nasogastric tube is in place, with the tip located in the stomach. 

Impression: The history, clinical data, and x-ray findings support the diagnosis of duodenal atresia.

This infant was taken to surgery for an exploratory laparotomy. Duodenal atresia was diagnosed, and an end-to-end anastomosis was performed.

Small Bowel Anomalies: Jejunal-Ileal Obstructions

Obstructions of the jejunum are most commonly the result of an intrauterine ischemic insult.13,14,19 Jejunal and ileal atresias are two times more common than is duodenal atresia, with ileal atresia being more common than jejunal atresia.15 The four major types of ileal atresia are shown in Figure 9.

Type I atresia shows the atretic portion of the lumen continuous with the dilated portion, but occluded by a thin-walled web. Type II shows discontinuity of the two ends, which are connected by a fibrous cord of atretic bowel. Type IIIa, the most common, is similar to Type II but without the connecting fibrous cord, and a V-shaped deficit of variable size is noted. Type IIIb, a subcategory of Type III referred to as the "apple peel" or "Christmas tree," is assigned when the V-shaped deficit is extremely large and there is agenesis of the dorsal mesentery. Type IV atresia occurs when there are multiple areas of atresia.13,14,18,19 Anomalies associated with jejunal and ileal atresias include malrotation, volvulus, gastroschisis, intussusception, and cystic fibrosis.15,19

Clinical Features

Polyhydramnios may or may not be a presenting sign of...
jejunal-ileal obstructions. It is seen more with proximal jejunal atresia than with distal atresia. Abdominal distention occurs in 75–80 percent of patients, along with bilious emesis, large gastric aspirates, no stool after the first 24 hours of age, history of poor feeding, and indirect hyperbilirubinemia.

**Radiographic Imaging**

The radiograph of an infant with proximal jejunal atresia may be diagnostic alone, showing a characteristic “triple bubble” (Figure 10). More distal obstructions, however, may show only multiple areas of dilated loops of bowel, requiring a contrast enema to identify the precise area of obstruction. Dilated loops of bowel that have an inverted U shape are characteristic of a small-bowel obstruction.

**Case Study**

Baby Girl C is a 36 week gestation, 3,240 gm female born by cesarean section to a 22-year-old gravida 1, para 0, Hispanic mother with good prenatal care. Maternal history: VDRL negative, rubella immune, HbsAg negative, and AFP (alpha-fetoprotein) normal. The pregnancy was uncomplicated, except for a fetal ultrasound at 32 weeks revealing multiple dilated loops of bowel with polyhydramnios. Spontaneous rupture of membranes occurred five hours prior to delivery. Upon delivery, the neonate was noted to have profound abdominal distention. A nasogastric tube was placed and 125 ml of bilious gastric aspirate were obtained. Apgar scores were 1, 5, and 8 at one, five and ten minutes, respectively. Baby Girl C had no respiratory effort and was immediately intubated and transferred to the NICU, where an additional 300 ml of bilious fluid were suctioned from the stomach. Fluid, resuscitation, and inotropic support for hypotension were required. A surgical consultation was obtained immediately.

Figure 11 shows this infant’s abdominal film.

**X-ray Evaluation (Figure 11)**

**Indication** for the x-ray was prenatal suspicion of GI obstruction, abdominal distention, and a large gastric aspirate. **Penetration** appears to be overexposed. **Rotation** is slightly to the left. The **soft tissues** of the neck, chest, and extremities are of normal thickness and without emphysema. The **body framework** is intact, with 12 ribs bilaterally and normal vertebrae. The **trachea** shows a straight air column, indicating an inspiratory film. The trachea bifurcation can be seen at T4. The endotracheal tube is at T3. The **hilum** appears of normal size, and the heart is normal in shape, size, and location. The **diaphragm** is at T8 on the right and T9 on the left. The **lung fields** show slight interstitial edema, with a minor fissure noted between T5 and T6. The endotracheal tube is at T3. An umbilical line is at T6 on the left side of the vertebral column. A central catheter is noted, with the tip in the left subclavian vein. The **abdomen and stomach**: Gastric air is present on the left, with numerous dilated loops of bowel. The tip of the nasogastric tube is in the stomach.

**Impression**: The history, clinical data, and x-ray findings support the diagnosis of intestinal obstruction.

After stabilization, this infant was taken to surgery for an exploratory laparotomy, which revealed a jejunal-ileal atresia. A small-bowel resection with primary anastomosis, omentectomy, appendectomy, and placement of jejunostomy tube was performed.
The Colon and Rectum

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The large bowel includes the cecum, colon, rectum and anal canal. The relative positions of these structures radiographically can be reviewed by referring to Figure 1. The ileocecal valve is proximal to the cecum and colon and prevents reflux of feces. The ascending colon is fixed retroperitoneally to the posterior abdominal wall. The transverse colon has a mesentery, the mesocolon, and is attached to the posterior surface of the omentum. The descending colon is fixed retroperitoneally and the sigmoid colon has a mesentery that permits mobility. The rectum is relatively straight in infancy and the lower rectum lies below the peritoneal reflection. The short anal canal contains the internal and external sphincters and is attached posteriorly to the sacrum. Two anomalies will be discussed here: one of the colon, Hirschsprung’s disease, and one of the rectum, imperforate anus.

Development of the Large Bowel

During early gestation, the hindgut is a straight and hollow tube. Rapid growth and insufficient space in the abdominal cavity for the developing bowel cause the midgut and hindgut to herniate into the umbilical cord around the sixth week of gestation and return to the abdomen during the tenth week.20,21 During this stage of development, numerous congenital anomalies may occur, including omphalocele, malrotation and volvulus, intestinal stenosis and atresia, and anorectal malformations.13,20 Functional disorders of the large bowel may also occur. These include meconium plug syndrome, small left colon, and Hirschsprung’s disease.

A thorough physical examination, history, and clinical presentation can help in determining a diagnosis. Significant findings may include (1) abdominal distention, (2) nonbilious and/or bilious emesis, (3) failure to pass meconium, and (4) diarrhea.13,21 Radiographic imaging using a flat plate film may help identify an obstruction, but the majority of large-bowel anomalies are diagnosed by contrast studies.13,21,22

Large-Bowel Anomaly: Hirschsprung’s Disease

Hirschsprung’s disease, or congenital aganglionic megacolon, is a common cause of bowel obstruction in the neonate.13,16 The overall incidence is 1 in 5,000 births, with males affected three to four times more often than females.13,16,21,22 A familial association has been reported, but the majority of cases are not inherited.16,23 The disease is caused by the absence of intrinsic ganglion cells in the myenteric (Auerbach) and submucosal (Meissner) plexuses of the bowel wall.13,16,21,23 The aganglionic segment frequently involves the rectum or rectosigmoid area but may extend to the proximal colon or, in rare cases, to total colon agangliosis.13,16,21,23

Clinical Features

Most Hirschsprung’s disease cases present in the neonatal period. Within 24 to 48 hours of age the newborn shows abdominal distention and failure to pass meconium. Other clinical signs include bilious emesis, constipation, diarrhea, and failure to thrive.16,21,22 Hirschsprung’s disease can be associated with other congenital anomalies, including imperforate anus, urinary tract abnormalities, cardiac defects (e.g., ventricular septal defect), seizure disorders, malrotation, long bone defects, and Down syndrome.13,21,23

Radiographic Imaging

Plain abdominal films in neonates who are suspected of having Hirschsprung’s disease usually show evidence of distal small-bowel and/or colonic obstruction with markedly dilated loops of bowel, with or without air in the rectum (Figure 12).13,16,21,22 Diagnosing Hirschsprung’s disease from a plain abdominal film is often impossible. It is also difficult to differentiate between the small bowel and large bowel because of massive dilation.16

A contrast enema is a more clear-cut diagnostic tool in Hirschsprung’s disease. With Hirschsprung’s disease, there is
usually a disparity in size between the small aganglionic distal bowel and the dilated proximal bowel. However, this transition zone is not always noted in early infancy, making the diagnosis more difficult. The rectosigmoid index (RSI) can be used in a contrast enema to indicate the presence of a transition zone. The RSI, the maximum diameter of the rectum compared with the maximum diameter of the sigmoid, is usually a 1:1 (or greater) relationship because the rectal diameter is normally larger. However, in Hirschsprung’s disease, the RSI is less than 1:1 because the rectal diameter is less than the sigmoid diameter (Figure 13).

The diagnosis is easier when there is a transition zone showing a sudden funnel with sharply angulated bowel walls. Also, in normal infants, most of the contrast material should be expelled within 24 hours; however, in infants with Hirschsprung’s disease, contrast material is usually retained, which is a reliable indicator (Figures 14 and 15).

The most definitive diagnosis of Hirschsprung’s disease is by rectal biopsy, however, which confirms the absence of ganglion cells.

**Case Study**

Baby Boy S, a 42-week, 2,865 gm infant, was born by emergency cesarean section, secondary to fetal distress, to a 34-year-old, gravida 5, para 3, African American mother. The...
pregnancy was reportedly uncomplicated. Maternal history was blood type O positive, VDRL negative, rubella immune, HbsAg negative, and HIV negative. Although the mother denied use of drugs during pregnancy, the infant’s toxicology screen was positive for cocaine. Upon delivery, the infant was active but required blow-by oxygen for cyanosis. Apgar scores were 8 and 9 at one and five minutes, respectively. Baby Boy S was taken to the newborn nursery. At 3 hours of age, he took his first feeding, and passage of meconium was noted at 4 hours of age. The infant tolerated subsequent feedings until approximately 14 hours of age, at which time he had a large emesis and was not interested in nipping. Around 24 hours of age, he was reported to be lethargic and refusing to eat, with abdominal distention and decreased bowel sounds. The initial KUB (plain film) showed multiple dilated loops of small bowel, but no air/fluid levels or free air. An abdominal film with contrast was then done (Figure 16).

Two saline enemas were given, with no resulting passage of stool, and the infant’s abdominal girth increased from 30.5 cm to 36 cm in two days. Baby Boy S was transferred to the NICU for surgical evaluation and management.

X-Ray Evaluation (Figure 16)

**Indication** for the x-ray was abdominal distention, failure to pass stool, and KUB showing multiple dilated loops of bowel. **Penetration:** appears to be slightly overpenetrated.

**Rotation:** appears to be slightly to the left. The **soft tissues** of the neck and chest are not visible. The soft tissues of the extremities are of normal thickness. The **body framework** is intact, with normal vertebrae and long bones of the legs. The **diaphragm** is at T8 on the right and T9 on the left. The **abdomen** is distended, with the stomach on the left, decompressed, with an orogastric tube in place. There are multiple dilated loops of small and large bowel, with contrast material in the rectum and colon. The rectum and rectosigmoid colon appear normal in size, with dilation of the transverse and ascending colon. Note the angular areas and serrated edges along the bowel wall. **Impression:** The history, clinical data, and x-ray findings support the diagnosis of Hirschsprung’s disease.

The infant was taken to surgery, where a segmental colon resection and colostomy were performed. During surgery, a narrow transition zone was observed distal to the ileocecal valve. Biopsies, taken from both the proximal ascending colon and the distal colon, revealed absence or near-absence of ganglion cells, consistent with the diagnosis of Hirschsprung’s disease.

**ANORECTAL MALFORMATION:** **IMPERFORATE ANUS**

Imperforate anus is defined as an arrest of rectal descent, resulting in an absence of an anal opening. There are two types: high and low imperforate anus. The classification of imperforate anus depends on the level to which the rectal
pouch has descended. The puborectalis sling levator muscle (responsible for fecal continence), the pubococcygeal line, and the ischium ("i") point are landmarks used to identify the position and type of the anal defect.³

In high imperforate anus, the rectum terminates above the sphincter muscle complex (supralevator) as well as the pubococcygeal line. The sphincter muscle complex consists of (1) the levator ani muscles, (2) the superficial external sphincter muscle, (3) the subcutaneous external sphincter muscle, and (4) the puborectalis muscle. In low imperforate anus, the rectum has descended below the sphincter muscle complex (translevator) and below the "i" point.³

Diagnosis of imperforate anus is made by physical examination of anal features and by radiographic imaging, such as a plain film of the abdomen and an invertogram. An abdominal plain film is taken to assess the presence of bowel gas in the rectum. The invertogram is done to determine the level of the blind rectal pouch.³,13,25 For the latter radiographic study, the infant is positioned upside-down for at least three minutes before the film is taken.³ A metal marker is taped to the skin in the area of the external sphincter to indicate the cutaneous level of the anus.

When the clinician is interpreting the invertogram, the pubococcygeal line and the ischium point are determined. If the air in the blind rectal pouch ends at or above the pubococcygeal line, the defect is considered high. If the air has penetrated below the ischium point, the defect is considered low.³,25

Surgical management of imperforate anus consists of a colostomy for high lesions and perineal anoplasty or dilatation for low lesions.³,24,25

Case Study
Baby Boy M, a 3,350 gm infant, was born at 42-weeks gestation by spontaneous vaginal delivery to a 29-year-old Hispanic mother, gravida 4, para 2, Ab 2. Maternal history was as follows: blood type O positive, VDRL nonreactive, HbsAg negative, rubella immune, and tuberculin purified protein (PPD) negative. Fetal ultrasound done early in pregnancy confirmed the mother's dates. The pregnancy as well as the labor and delivery were uncomplicated. Infant Apgar scores were 8 and 9 at one and five minutes, respectively. Resuscitation consisted of blow-by oxygen and vigorous stimulation. The infant responded well. During physical examination in the delivery room, the infant was noted to have a bilateral cleft lip and palate and imperforate anus.

Baby Boy M was transferred to the NICU for further evaluation and management. An abdominal plain film taken at two hours of age revealed an absence of bowel gas in the rectum (Figure 17). Subsequently, an invertogram was taken (Figure 18).

X-Ray Evaluation (Figure 18)
The indication for the invertogram was to identify the level of the blind rectal pouch.

Penetration: appears normal.
Rectum: Blind rectal pouch penetrates below the ischium point.
Impression: The history, clinical presentation, and radiographic findings are consistent with low imperforate anus.

After the diagnosis was confirmed during surgery, an anoplasty was performed.

REFERENCES